Journal of
Cardiology & Cardiovascular
Surgery

Case Report

DOI: 10.51271/JCCVS-0039

Mitral annular disjunction as the cause of malignant ventricular arrhythmia in a young adult: a case report

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Cite this article: Perkovic A, Chuah Eunice, Mooney J. Mitral annular disjunction as the cause of malignant ventricular arrhythmia in a young adult: a case report. *J Cardiol Cardiovasc Surg.* 2024;2(3):62-64

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Received: 25/04/2024 ◆ **Accepted**: 22/07/2024 ◆ **Published**: 30/09/2024

ABSTRACT

Mitral annular disjunction (MAD) is associated with ventricular arrhythmias and sudden cardiac death (VA/SCD). Risk prediction for VA/SCD in individuals with MAD includes clinical, ECG and imaging markers. Imaging assessment for MAD includes transthoracic echocardiography (TTE) and cardiac magnetic resonance imaging (CMRI) and there are certain features in each modality to aid both diagnosis and risk assessment. A 23-year-old female with no medical history presented to hospital following out of hospital cardiac arrest. Effective bystander CPR and successful cardioversion from ventricular fibrillation to sinus rhythm by paramedics achieved return of spontaneous circulation. Focused transthoracic echocardiography and cardiac magnetic resonance imaging identified MAD. Notably, there were no specific high risk clinical or imaging features. The patient recovered completely and was discharged with implantable defibrillator for secondary prevention. MAD is a rare cause of VA/SCD. Arrhythmias secondary to MAD are more likely with abnormalities on cardiac imaging, including late gadolinium enhancement, fibrosis of the mitral annulus or papillary muscles, mitral valve pathology and ventricular ectopic beats. In this case, none of the traditional risk factors for VA/SCD were present. Thus, MAD remains an important differential in all patients with an otherwise unexplained cardiac arrest, even if high risk features are not present.

Keywords: Primary cardiac tumor, rhabdomyosarcoma, cardiooncology

INTRODUCTION

Mitral annular disjunction (MAD) is a structural cardiac defect that is identified by separation of the mitral annulus and ventricular myocardium in systole. It is commonly associated with mitral valve prolapse (MVP) and resultant mitral regurgitation (MR).[1] The prevalence of MVP in the general population is 2-3%, and a sizeable minority (estimated 30%) of these have associated MAD. Conversely, most but not all patients with MAD (78%) can have concurrent MVP.[2,3]

MAD is associated with ventricular arrhythmias and sudden cardiac death (VA/SCD),[2] and these can occur irrespective of whether MVP is present or absent.[3] Changes in mechanical function of the valvular and annular apparatus, and subsequent tissue changes including regional fibrosis are thought to be the source of VA/SCD.[1] Features associated with higher risk of VA/SCD in MAD include recurrent palpitations, ECG and telemetry changes, longer disjunction distance, and presence of papillary muscle fibrosis on cardiac magnetic resonance (CMR).[2-4]

We present a case of a young adult whose sentinel presentation was aborted cardiac arrest, with hallmark features for MAD on multimodality imaging. Of interest, our case lacked high risk clinical and imaging features for VA/SCD. This case highlights both the heterogeneity of presentation, and the utility of multimodality imaging in conjunction with exclusion of alternative causes for SCD.

CASE

A 23-year-old Caucasian female with no known medical history presented to our hospital following cardiac arrest at work while cleaning windows. A colleague commenced effective bystander cardiopulmonary resuscitation for approximately 10 minutes prior to arrival of paramedics. Initial rhythm was ventricular fibrillation and a single 200 joule shock resulted in successful reversion to sinus rhythm.

A detailed history revealed 2 previous episodes of syncope without presentation to a medical care provider and infrequent, brief palpitations in the preceding 5 years. There was no known family history of cardiomyopathy or SCD. She denied any recreational drug use or excessive exercise. Physical examination was normal.

Electrocardiography (ECG) showed an incomplete right bundle branch block (RBBB) with QRS interval of 112ms. Corrected QT interval was 410 milliseconds. Continuous

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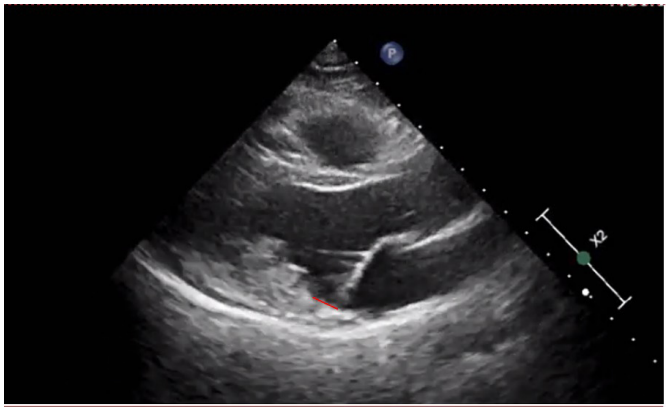

Figure 1. Parasternal long axis window at end systole with MAD distance of 8 mm.

telemetry monitoring did not reveal arrhythmia or significant pauses and intravenous flecainide challenge and high precordial lead ECG did not elicit Bragada pattern ECG changes. Initial bloods were unremarkable apart from mild hypokalaemia (3.1mmol/L) and hyperlactatemia (4.0mmol/L).

Initial transthoracic echocardiography (TTE) showed normal left ventricular size, systolic function and ejection fraction. No valvular or annular pathology was identified apart from trivial MR, and no leaflet thickening. A subsequent focused TTE at a tertiary institution demonstrated MAD, with end systolic distance of 8mm in the posterolateral window.[5] There was minimal leaflet prolapse and trivial MR with slightly posteriorly directed jet (Figure 1).

CMR at our hospital demonstrated 6.6 mm annular disjunction in 3 chamber sequence on the posterior wall (red line; figure 2), with 4mm atrial excursion of the mitral valve leaflets at end systole (blue lines; figure 2). Disjunction was also evident at the inferior wall. There was no myocardial or papillary late gadolinium enhancement (LGE) evident. Posterior wall myocardial 'curling motion' was seen. No other structural deficits were evident, and chamber size and function were in normal ranges. Coronary computed

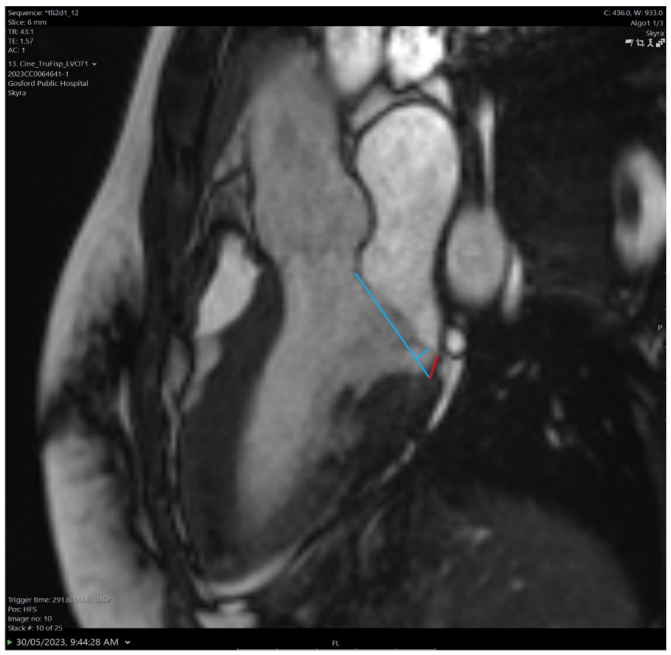

Figure 2. Parasternal long axis sequence, cardiac MRI. Longitudinal MAD distance of 6 mm (red line), with 5 mm atrial excursion of mitral annulus (blue lines).

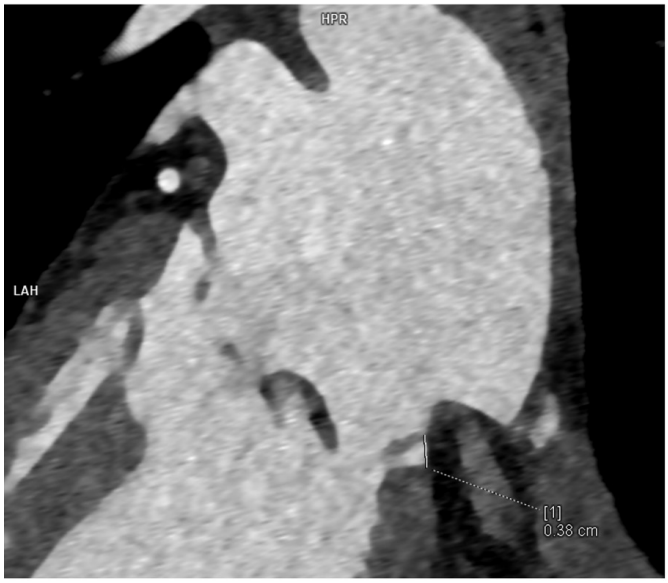

Figure 3. CCTA
Mid diastolic images, with disjunction noted at inferior wall between P1 and P2 scallops of posterior mitral valve leaflet.

tomography angiography (CCTA) acquired in mid diastole demonstrated normal coronary vessels with normal origins. Annular disjunction was again evident (Figure 3).

The patient was transferred to a tertiary institution where exercise stress testing and screening for metanephros's and catecholamines were unremarkable. Implantable cardioverter-defibrillator was inserted for secondary prevention. Genetic testing was not performed as testing to elicit changes consistent with Bragada syndrome and other channelopathies were unremarkable, and an alternative cause for SCD was found.[6]

The patient was discharged home, and at 3 month follow up remained well with no further events or recorded arrhythmias.

DISCUSSION

We present a case of aborted cardiac arrest in a young adult with characteristic imaging features for MAD, and no alternative causes for VA/SCD. In comparison to known high risk features in MAD, our case lacked a number of these. Our patient was significantly younger, with first presentation as cardiac arrest. There were concerning features in the history preceding this admission which were only attained in retrospect, and there was no pre-hospital evidence of arrhythmia or structural heart disease. In addition, there were no high-risk features on the subsequent imaging. Regardless, our case reflects recent studies,[3,4] where the presence of MAD alone can be a high-risk marker for arrhythmic events.

Our case demonstrates some of the utility and limitations of different imaging modalities to diagnose and risk stratify MAD. Firstly, initial TTE at our institution did not identify MAD, and this may have been due to the lack of associated features including MV prolapse or MR to highlight suspicion. Though an initial screening tool, the sensitivity of TTE for MAD is modest, with estimates of approximately 65%,[7] which reflects our experience and highlights the value of integrated imaging to improve diagnostic accuracy. Second, CMR which is considered to have higher sensitivity[4] identified MAD and associated annular displacement and myocardial curling.

This reflects the utility of CMR for the diagnosis of MAD as a gold standard.[1] Papillary fibrosis was not present in our case, however in a high-risk cohort it was only evident in 36% of cases,[3] and this may reflect our patient's young age and lack of time to develop fibrotic changes and other features of mitral dysfunction. The presence of disjunction on the inferolateral wall, only seen in approximately 5% of cases of MAD,[8] may suggest that this feature is associated with VA/SCD. Thirdly, MDCT and subsequent TTE helped confirm the diagnosis, and these reflect both the high specificity of TTE[7] and the potential use of CCTA.[9] Finally, advanced imaging markers including tissue tracking and extracellular volume may have greater predictive utility for diagnosis and stratifying risk,[10] though were not available for this case and remain the subject of ongoing investigation.

A 2022 European heart rhythm association consensus guideline acknowledged that, based on available literature, MAD in the absence of MVP has an unclear association with significant outcomes.[1]

CONCLUSION

This report reinforces the notion that the presentation of MAD is widely heterogenous and should be considered in all individuals with unexplained cardiac arrest, regardless of the presence of MVP. Additionally, risk prediction in these individuals remains unrefined and would otherwise not have predicted VA/SCD in this patient. Ongoing utilization of cardiac imaging markers may further stratify risk in these patients.

ETHICAL DECLARATIONS

Informed Consent

All patients signed and free and informed consent form.

Referee Evaluation Process

Externally peer-reviewed.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

Financial Disclosure

The authors declared that this study has received no financial support.

Author Contributions

All of the authors declare that they have all participated in the design, execution, and analysis of the paper, and that they have approved the final version.

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